

## Progressive scoliosis in early, non-progressive CNS injuries: role of axial muscles

JOHN C. DETOLEDO<sup>†</sup> and  
HELENA HADDAD<sup>‡</sup>

<sup>†</sup>Department of Neurology, University of Miami, Miami, FL, USA.

<sup>‡</sup>Fairview Training Center, Salem, Oregon, USA

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Forty-three patients with progressive neurological deficits involving axial musculature, starting 3-6 years after non-progressive brain injuries insults are described. Losses of function followed period of several years of stable motor deficits. Subsequent losses were stereotypic, with loss of ambulation and scoliosis, followed by loss of word articulation, malalignment of the mandible and ultimately neurogenic impairment of swallowing. Physical therapy, serial castings and spinal instrumentation palliated specific musculoskeletal problems but did not alter the relentless loss of various functions. The balanced action of paired axial muscles (i.e. spine, proximal muscle groups of the lower extremities, oropharynx, mastication) is regulated by the brainstem with modulation by the cerebral hemispheres. The clinical evolution in these patients suggest that, in the absence of normal input from the cerebral hemispheres, some patients have a progressive loss of these brainstem mechanisms. The most resistant functions (last ones to be lost), seem to be the ones phylogenetically most relevant for survival, such as suction and swallowing.

### Introduction

The association of severe musculoskeletal problems with spastic disorders of childhood is well described in the literature [1-3]. With few exceptions, these reports examine each problem in isolation, usually from the orthopaedic, neurological or rehabilitation points of view. Each of these deficits however, are not isolated events, and represent only one aspect of a much broader problem due to a chronically malfunctioning CNS. This paper reports a pattern of progression of skeletal deformities and loss of neurological functions after a non-progressive perinatal injury, that evolves over the first two decades of life in a population of institutionalized, developmentally disabled patients [4, 5].

### Materials and methods

The neurologic and orthopaedic deficits in a population of 624 institutionalized developmentally disabled patients were reviewed. All patients had lived in the institution all their lives. Medical records, including physical therapy notes and radiology reports dating back to birth, were available for most of them. The records were reviewed with attention to age of CNS insult, aetiology of MR, neurological,

orthopaedic and physical therapy reports of patient's evolution over the years. All patients were examined by both authors at the institution over a 4-year period. Changes in patients' status were obtained by comparing the serial examination from the records and also from interviewing family members and staff who knew the patients.

### Findings

Of the 624 patients, 43 patients had a similar clinical and temporal progression of musculoskeletal problems (table 1). These patients are the subject of this report. All 43 patients were bedridden, 36 were profoundly retarded, seven had moderately severe retardation, 20 were male, 23 female; mean age was 27 (range 18–38 years). Twenty-six patients had a history of perinatal hypoxia-ischemia, six had a history of CNS infection, one had tuberous sclerosis, the aetiology of mental retardation was undetermined in 10 patients. After the perinatal injury, patients had a 3–7 year period of apparently stable motor deficits. Twenty-eight patients had a history of seizures, none had a discernable neuromuscular disorder.

The neurological deficits are summarized in table 2. Scoliosis, often with multiple curvatures and various degrees of rotation, was present in all patients. Scoliosis was first noted before the age of 6 years in 38 of the patients, onset could not be ascertained in five. Scoliosis had a slowly progressive course initially, with rapid progression with growth in the early teens. Seven patients developed full collapsing curves despite continued physical therapy and external bracing. Early assessments reported osteoporosis in 32 patients and pelvic obliquity in 13 patients.

Contractures of soft tissues and skeletal deformities of the extremities were present in 37 patients, usually in combination with severe spasticity. Both proximal and distal muscle groups of the extremities were affected. Deformities of the hands and feet followed no specific pattern (i.e. flexors versus extensors). Contractures of the lower extremities were mostly in extension at the knee with spastic adduction of the hip. Twenty-one patients had previous serial castings for equinus tightness. Seven patients had a combination of plantar flexion contracture in one foot and extension contracture on the other. Hand contractures were usually in flexion with either ulnar or radial deviation. Upper and lower extremities were always involved bilaterally although the severity of the involvement was often asymmetric. None of

Table 1. Age of onset of CNS insult and progression of deficits in the 43 patients

Age of onset (years)	Initial CNS insult	Onset of decline after stable	Scoliosis first detected (38 pts)	Loss of word articulation (9 pts)	Swallowing difficulties (36 pts)
< 2	33	4	4?	—	—
< 4	4	7	7	—	—
< 6	—	12	21	2?	—
< 8	—	7	—	2	—
< 10	—	2	—	1	11
< 15	—	—	—	4	19
< 20	—	—	—	—	4
Unknown	6	11	6	—	—
Total	43	43	38	9	36

Table 2. Neurological findings in the 43 patients

Severe mental retardation	36
History of seizures	28
Progressive kyphoscoliosis	43
Fixed deformities of extremities	37
Distal deformities > proximal deformities	33
Strictly unilateral neurologic dysfunction	0
Asymmetries on neurologic exam	40
Hyperactive masseter reflexes	31
Gag reflex present	33
Macroglossia	17
Limitations in mandibular ranges of motion	36
Previously able to communicate verbally	9

the patients had evidence of a strictly unilateral dysfunction. Restriction of ranges of motion of proximal muscle groups was more common in the lower extremities than in the uppers. Nine patients had been previously able to communicate verbally, albeit at a limited level. Whereas comprehension seemed not to have changed much, the ability to communicate verbally was gradually lost and none of the nine patients had any meaningful verbal communication left by their early teens.

Thirty-six patients had progressive swallowing difficulties requiring gastrostomy. Gastrostomies were performed for both nutrition and airway protection purposes. These patients were the most severely impaired. The mean age of feeding tube placement was 21.1 (15-31 years) while the mean age of the 43 patients was 27.1 years. Six patients underwent internal spinal fixation with Harrington and Luque instrumentation. Despite the stabilization of the kyphoscoliosis after the surgery, contractures of the extremities, mandibular malalignment and swallowing difficulties continued to deteriorate. Mandibular ranges of motion, assessed by maximal jaw opening and lateral jaw excursion were limited in all patients.

## Discussion

Perinatal CNS insults are commonly thought of as causing non-progressive cerebral motor deficits [6]. This, however, is not invariably the case, and deterioration of the clinical deficits have been reported under certain circumstances [3, 7].

The chronic problems seen in these 43 patients with early CNS injuries followed a relatively stereotypic pattern. In addition to the neuromuscular scoliosis, there was progressive loss of function of other paired axial muscles over 1-2 decades. This slowly progressive course cannot be explained on the basis of the acute, non-progressive loss of neurons occurring at birth in these 43 patients. A subsequent chronic loss of neurons, either by loss of trophic influences [8, 9] or reduced numbers of surviving neurons [10], are more likely explanations.

The innervation of paired axial muscles in under dual CNS control, one mediated by supratentorial, volitional mechanisms, the other, largely reflex in nature, originates from subcortical and brainstem structures [11-17]. The medial subcortical system originating in the tectum, interstitial nucleus of Cajal and medial reticular formation in the brainstem, is concerned with the reflex innervation of axial structures [16, 18]. This system modulates tone and synergy of the paired muscles. Experimental lesions of the medial system in primates result in persisting

postural instability due to impaired function of the axial musculature and proximal muscles of the extremities [16]. Cortical input greatly modifies the severity of the deficits following lesions of the medial system. Anatomically identical lesions in the medial system will produce more severe deficits in animals given hemispheric lesions [16].

The alignment of the spine is determined, in part, by the balanced action of paired axial muscles, the paraspinals and obliquii abdominalis, a mechanism proposed almost 250 years ago [19]. These muscles have complex interactions integrated by the brainstem and spinal cord, being synergistic or antagonistic depending on the type of movement. Flexion and extension movements of the spine, for example, are accomplished by the synergistic action of both paraspinal muscles, whereas lateral tilting or rotational spinal movements, contract muscles on one side while the ones on the opposite side relax. Some proximal muscle groups of the lower extremities, the cervical musculature and the facial and oropharyngeal muscles responsible for phonation and deglutition are also axial in location and paired in function. Thirty-six of the 43 patients, clinically the more severely compromised ones, also developed dysfunction of the paired musculature of the oropharynx with loss of articulation of words and swallowing difficulties [20], both regulated by brainstem mechanisms.

The cluster of symptoms in this series was similar in patients with different types of CNS insults suggesting that the aetiology of the lesions was not the determining factor for the appearance or progression of the symptoms. Patients with more severe insults and initially more severely compromised (i.e. never ambulated or acquired language) exhibited signs of progressive deterioration earlier than less impaired patients. Clinically, all patients had signs and symptoms of diffuse CNS dysfunction including brainstem structures, none had strictly unilateral findings although asymmetries were present in 18 of the 43 patients.

The term 'static encephalopathy' is often used in patients with developmental disabilities following early CNS injuries. This label is misleading as some patients have a progressive, almost malignant decline after an initial period of apparent stability, that progresses over 10–20 years. In these patients the musculoskeletal deformities are not isolated events, and represent only one aspect of a much broader problem due to a chronic CNS dysfunction. The salient feature common to these patients was a severe, diffuse, CNS injury occurring early in life. Clinically, patients presented with severe spasticity and progressive, bilateral loss of previously stable motor functions over the course of years. Loss of ambulation, neuromuscular scoliosis with multiple curvatures, contractures of soft tissue, anarthria and dysphagia were present in most patients.

The presence of this cluster of symptoms in the first decade of life anticipates a poor overall prognosis. Spinal curvatures typically progress to extreme degrees despite physical therapy and castings. Surgical instrumentation of the spine arrests the scoliosis but not the overall decline of these patients.

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